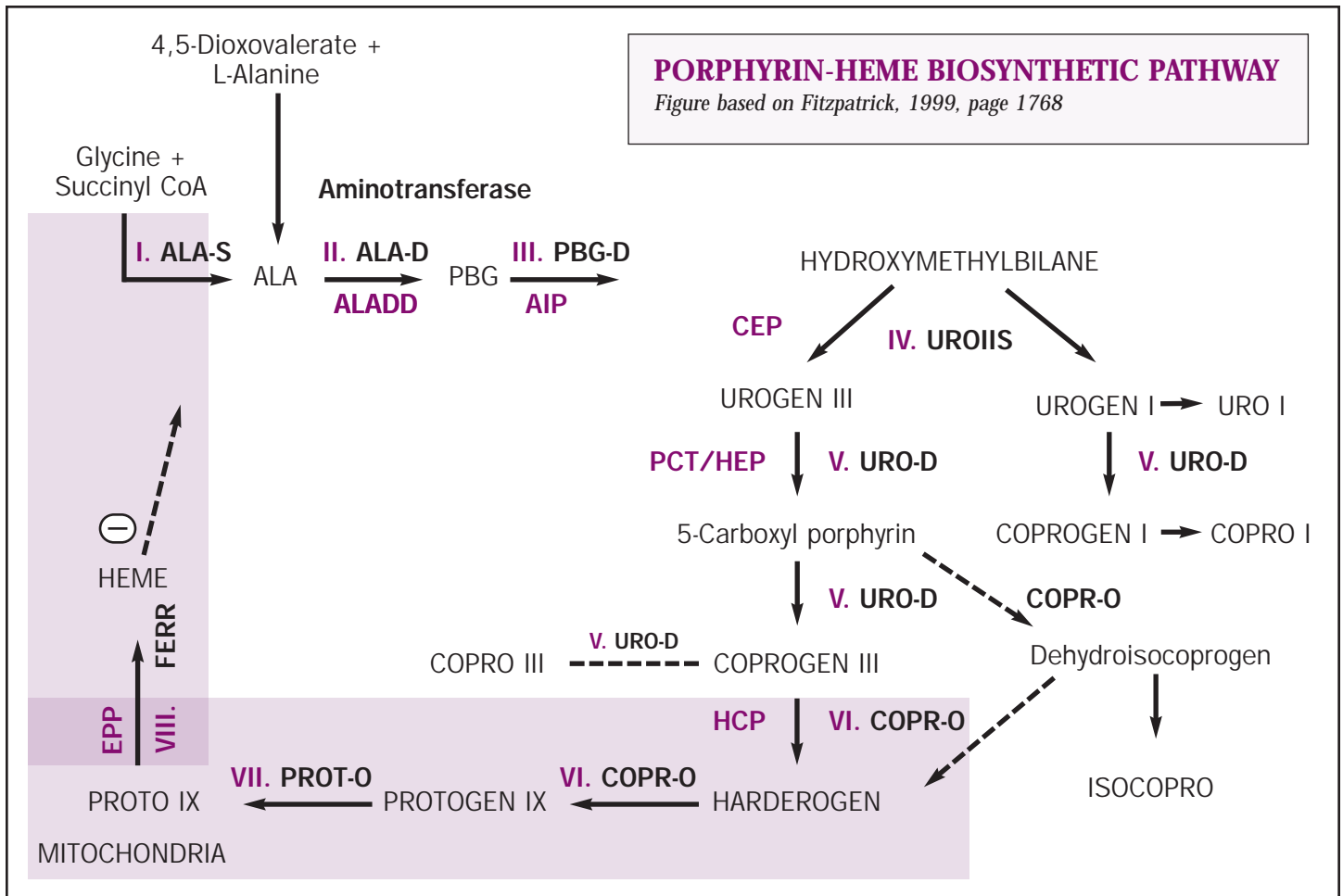


Boards' Fodder: Porphyrrias

Sharon E. Jacob, M.D., and Hari Nadiminti, B.S.



KEY:

Roman numerals represent chronological order, starting with **ALA-s** (rate limiting step enzyme)

Diseases in dark purple, enzymes in bold. Light purple indicates mitochondria

MNEUMONICS:

4 mitochondrial enzymes: "ALAS, FERRous OXIDizes" [ALAS, Ferrochelatase & the Oxidases]

AIP & **ALADD**: Absent skin findings

HCP, **AIP**, and **VP**: **HAVE** acute attacks of ALA, PBG

"No pee pee in EPP" [no porphyrins in the urine in EPP]

ABBREVIATIONS:

- | | |
|--|---|
| ALA-S : aminolevulinic acid synthase | HCP : hepatic coproporphyrin |
| ALADD : ALA dehydratase deficiency | HEP : hepatic erythropoietic porphyria |
| AIP : acute intermittent porphyria | PBG-D : porphobilinogen deaminase |
| ALA-D : ALA dehydratase | PCT : porphyria cutanea tarda |
| CEP : congenital erythropoietic porphyria | PROT-O : protoporphyrin; |
| COPR-O : corprotoporphrin oxidase | PROTOGEN : protoporphyrinogen |
| COPROGEN : coproporphyrinogen | URO : uroporphyrin |
| EPP : erythropoietic protoporphyria | UROGEN : uroporphyrinogen |
| FERR : ferrochelatase | URO-D : urogen decarboxylase |
| HARDEROGEN : harderoporphyrinogen | UROIIIIS : urogen synthase III |
| | VP : variegate porphyria |



SHARON E. JACOB, MD.

Sharon E. Jacob, M.D., is assistant professor of clinical dermatology, director of medical education and contact dermatitis, department of dermatology and cutaneous surgery, University of Miami School of Medicine, Miami, Florida.

	ENZYME DEF.	DISEASE	INHERITANCE-	CLINICAL FEATURES	TREATMENT	RBC	URINE	STOOL	FLURO
I	ALAS					Proto	ALA/ Copro	ALA	
II	ALA-D	ALADD	A ^R	Rare (< 10 reported cases); sxs can mimic AIP & are highly variable – failure to thrive in infant & polyneuropathy in a 63 yo; r/o exposure to styrene (inhibitor of ALAD)	Acute attacks: Hematin				
		Tyrosinemia		Can mimic ALADD, b/c pts with hereditary tyrosinemia accumulate succinylacetone (inhibitor of ALAD)	Diet to minimize the phenylalanine-tyrosine				
III	PBG-D	AIP	A ^D	Incidence: 5 / 100,000; women > men (2:1); onset: 18 -40 yo; sx sequence: abd colic -> psychiatric sxs, ie hysteria -> peripheral neuropathy; NO SKIN FINDINGS; SIADH -> hyponatremia; urine discoloration; risk Hepatic CA	Glucose load, Hematin/cimetidine Pain: narcotics; Liver transplant – cure (1 case report)	PBG deami- nase	ALA, PBG Watson- Scharwtz test	N/ ALA/PBG in attacks	
IV	UROIIIS	CEP (Gunther's)	A ^R	Rare (<200 reported cases); onset: infancy: marked photosensitivity (vesiculo-bullous-scarring), increased fragility and ulcers lead to scarring; "werewolf-facies"; hypertrichosis, erythrodontia; hemolytic anemia; splenomegaly; port wine urine; corneal scarring -> blindness; acro-osteolysis; contractures	Sun avoidance, Splenectomy, BMT, β-carotene, transfusions, alphatocopherol	Uro > Copro	Uro > Copro	Copro > Uro	Teeth Urine, RBC
V	URO-D	PCT	A ^D Types: I: Sporadic II: Familial	Most common porphyria; onset: middle age; moderate photosensitivity, fragility of sun-exposed skin after trauma -> erosions & bullae-> scars, hyper/hypopigmentation, milia; hypertrichosis; scarring alopecia; photo-oncholysis; sclerodermoid plaques; dystrophic calcifications; serum iron normal; DM 25%; liver iron overload; mutation: HFE C2a2	EtOH elimination, sun protection, Phlebotomy to Hb<10, antimalarials	N	Uro > copro	Isocopro	Urine
		HEP	A ^R	Homozygous form of PCT; URO-D < 10%; onset: infancy; extreme photosensitivity; similar sxs as PCT (earlier onset)	Strict sun avoidance, Phlebotomy ineffective	Proto	Uro	Isocopro	Teeth
VI	COPR-O	HCP	A ^D	Incidence: 1-4 / 1,000,000; onset: 18-40; similar sxs to AIP, but less severe; 30% have skin findings: Hematinphotosensitivity -> blistering, scarring; during attach urine pink/red; mutation in CPOX	Glucose load, Hematin Pain: narcotics	N	Copro	Copro	
		Harderoporphyria		Rare form of HCP; onset: infancy; sxs similar to HCP, with jaundice and anemia					
		ECP		Rare (3 reported cases); mild skin photosensitivity		Copro		Copro	
VII	PROT-O	VP	A ^D	Common in South Africans; 15-30 yo; cClinically similar to AIP (abd colic, paralysis, psychosis) + PCT skin findings (photosensitivity); mutation PPOX; 624-626nm band	Glucose load, Hematin	N	Copro > Uro*	Proto > Copro	RBC
VIII	FERR	EPP	A ^D (A ^R)	Photosensitivity beginning in first decade; burning and tingling (non pruritic); edematous plaques-purpura, waxy scars; pox-like scarring on nose & cheeks; circumoral linear scars; weather-beaten skin; cholelithiasis; hepatic failure	Sun avoidance, β-carotene, Cholestyramine	Proto	N	Proto	